# DBT gene

dihydrolipoamide branched chain transacylase E2

## Normal Function

The *DBT* gene provides instructions for making part of an enzyme complex (a group of enzymes that work together) called branched-chain alpha-keto acid dehydrogenase, or BCKD. Specifically, the protein produced from the *DBT* gene forms a critical part of the enzyme complex called the E2 component.

The BCKD enzyme complex is responsible for one step in the normal breakdown of three protein building blocks (amino acids). These amino acids—leucine, isoleucine, and valine—are obtained from the diet. They are present in many kinds of food, particularly protein-rich foods such as milk, meat, and eggs. The BCKD enzyme complex is active in mitochondria, which are specialized structures inside cells that serve as energy-producing centers. The breakdown of leucine, isoleucine, and valine produces molecules that can be used for energy.

## Health Conditions Related to Genetic Changes

maple syrup urine disease

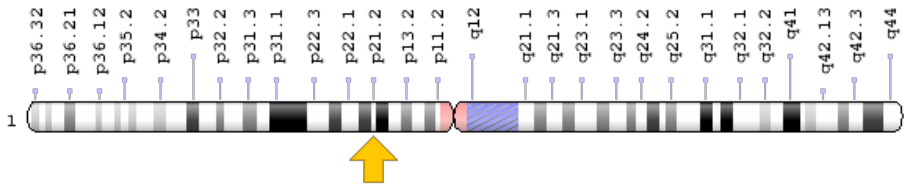
More than 30 mutations in the *DBT* gene have been identified in people with maple syrup urine disease, most often in individuals with mild variants of the disorder. Mutations in the *DBT* gene include changes in single DNA building blocks (base pairs) and insertions or deletions of a small amount of DNA in the *DBT* gene.

Mutations in the *DBT* gene disrupt the normal function of the E2 component, preventing the BCKD enzyme complex from effectively breaking down leucine, isoleucine, and valine. As a result, these amino acids and their byproducts build up in the body. This accumulation is toxic to cells and tissues, particularly in the nervous system. The buildup of these substances can lead to seizures, developmental delay, and the other medical problems associated with maple syrup urine disease.

## Chromosomal Location

Cytogenetic Location: 1p21.2, which is the short (p) arm of chromosome 1 at position 21.2

Molecular Location: base pairs 100,186,922 to 100,249,864 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)




Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- BCATE2
- dihydrolipoamide branched chain transacylase (E2 component of branched chain keto acid dehydrogenase complex; maple syrup urine disease)
- E2 component of branched chain keto acid dehydrogenase complex
- MSUD2
- ODB2_HUMAN

## Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1998): Major pathways of branched-chain amino acid metabolism (figure) https://www.ncbi.nlm.nih.gov/books/NBK20436/figure/A3097/
- Basic Neurochemistry (sixth edition, 1998): Maple syrup urine disease was the first congenital defect of branched-chain amino acid catabolism to be described https://www.ncbi.nlm.nih.gov/books/NBK28225/#A3107

GeneReviews

- Maple Syrup Urine Disease https://www.ncbi.nlm.nih.gov/books/NBK1319

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DBT+AND+maple+syrup+urine+disease%5BTIAB%5D%29+OR+%28dihydrolipoamide+branched+chain+transacylase%5BTIAB%5D%29%29+OR+%28%28dihydrolipoamide+branched+chain+transacylase%5BTIAB%5D%29+OR+%28MSUD2%5BTIAB%5D%29+OR+%28E2+%5Btiab%5D+AND+BCKD+%5Btiab%5D%29+OR+%28E2+%5Btiab%5D+AND+branched-chain+alpha-ketoacid+dehydrogenase+complex+%5Btiab%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- DIHYDROLIPOAMIDE BRANCHED-CHAIN TRANSACYLASE
  http://omim.org/entry/248610

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DBT%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2698
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1629
- UniProt
  http://www.uniprot.org/uniprot/P11182

## Sources for This Summary


- Chi CS, Tsai CR, Chen LH, Lee HF, Mak BS, Yang SH, Wang TY, Shu SG, Chen CH. Maple syrup urine disease in the Austronesian aboriginal tribe Paiwan of Taiwan: a novel DBT (E2) gene 4.7 kb founder deletion caused by a nonhomologous recombination between LINE-1 and Alu and the carrier-frequency determination. Eur J Hum Genet. 2003 Dec;11(12):931-6.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/14508502
- Flaschker N, Feyen O, Fend S, Simon E, Schadewaldt P, Wendel U. Description of the mutations in 15 subjects with variant forms of maple syrup urine disease. J Inherit Metab Dis. 2007 Nov;30(6):903-9. Epub 2007 Oct 8.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/17922217
- Henneke M, Flaschker N, Helbling C, Müller M, Schadewaldt P, Gärtner J, Wendel U. Identification of twelve novel mutations in patients with classic and variant forms of maple syrup urine disease. Hum Mutat. 2003 Nov;22(5):417.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/14517957

- Nellis MM, Danner DJ. Gene preference in maple syrup urine disease. Am J Hum Genet. 2001 Jan; 68(1):232-7. Epub 2000 Dec 7.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/11112664
  *Free article on PubMed Central:* https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1234918/
- Quental S, Macedo-Ribeiro S, Matos R, Vilarinho L, Martins E, Teles EL, Rodrigues E, Diogo L, Garcia P, Eusébio F, Gaspar A, Sequeira S, Furtado F, Lança I, Amorim A, Prata MJ. Molecular and structural analyses of maple syrup urine disease and identification of a founder mutation in a Portuguese Gypsy community. Mol Genet Metab. 2008 Jun;94(2):148-56. doi: 10.1016/j.ymgme.2008.02.008. Epub 2008 Apr 2.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/18378174
- Rodríguez-Pombo P, Navarrete R, Merinero B, Gómez-Puertas P, Ugarte M. Mutational spectrum of maple syrup urine disease in Spain. Hum Mutat. 2006 Jul;27(7):715.
  *Citation on PubMed:* https://www.ncbi.nlm.nih.gov/pubmed/16786533


---


Reprinted from Genetics Home Reference:
https://ghr.nlm.nih.gov/gene/DBT

Reviewed: December 2008
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services